

GEPHE SUMMARY

<p>tyrosinase-related protein 1 (TYRP1) (https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=^tyrosinase-related+protein+1+(TYRP1)^#gephebase-summary-title)</p> <p>Published</p>	<p>Gephebase Gene</p> <p>Entry Status</p>	<p>GP00002236</p> <p>Martin</p>	<p>GepheID</p> <p>Main curator</p>
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PHENOTYPIC CHANGE

<p>Morphology (https://www.gephebase.org/search-criteria?/and+Trait+Category=^Morphology^#gephebase-summary-title)</p> <p>Coloration (coat) (https://www.gephebase.org/search-criteria?/and+Trait=^Coloration+(coat)^#gephebase-summary-title)</p> <p>Valais Blacknecked goat ; black forequarters and white hindquarters</p> <p>brown instead of black forequarters in Coppernecked goat</p> <p>Taxon A</p> <p>Domesticated (https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=^Domesticated^#gephebase-summary-title)</p>	<p>Trait Category</p> <p>Trait</p> <p>Trait State in Taxon A</p> <p>Trait State in Taxon B</p> <p>Ancestral State</p> <p>Taxonomic Status</p>
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Taxon A	Latin Name	Taxon B	Latin Name
Capra hircus (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Capra+hircus^#gephebase-summary-title)	Capra hircus (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Capra+hircus^#gephebase-summary-title)	Capra hircus (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Capra+hircus^#gephebase-summary-title)	Capra hircus (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Capra+hircus^#gephebase-summary-title)
Common Name	Common Name	Common Name	Common Name
goat	goat	goat	goat
Synonyms	Synonyms	Synonyms	Synonyms
Capra aegagrus hircus; goat; domestic goat; goats; Carpa hircus; South African angora goat species	Capra aegagrus hircus; goat; domestic goat; goats; Carpa hircus; South African angora goat species	Capra aegagrus hircus; goat; domestic goat; goats; Carpa hircus; South African angora goat species	Capra aegagrus hircus; goat; domestic goat; goats; Carpa hircus; South African angora goat species
Rank	Rank	Rank	Rank
species	species	species	species
Lineage	Lineage	Lineage	Lineage
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Laurasiatheria; Cetartiodactyla; Ruminantia; Pecora; Bovidae; Caprinae; Capra	cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Laurasiatheria; Cetartiodactyla; Ruminantia; Pecora; Bovidae; Caprinae; Capra	cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Laurasiatheria; Cetartiodactyla; Ruminantia; Pecora; Bovidae; Caprinae; Capra	cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Laurasiatheria; Cetartiodactyla; Ruminantia; Pecora; Bovidae; Caprinae; Capra
Parent	Parent	Parent	Parent
Capra () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9922)	Capra () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9922)	Capra () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9922)	Capra () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9922)
NCBI Taxonomy ID	NCBI Taxonomy ID	NCBI Taxonomy ID	NCBI Taxonomy ID
9925 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9925)	9925 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9925)	9925 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9925)	9925 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9925)
is Taxon A an Intraspecies?	is Taxon A an Intraspecies?	is Taxon B an Intraspecies?	is Taxon B an Intraspecies?
No	No	No	No

GENOTYPIC CHANGE

<p>Tyrp1</p> <p>b; isa; Oca3; TRP1; Tyrp; TRP-1; brown; Tyrp-1</p> <p>10090.ENSMUSP00000006151 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000006151)</p> <p>Belongs to the tyrosinase family.</p>	<p>Generic Gene Name</p> <p>Synonyms</p> <p>String</p> <p>Sequence Similarities</p> <p>GO - Molecular Function</p>	<p>P07147 (http://www.uniprot.org/uniprot/P07147)</p> <p>()</p> <p>UniProtKB Mus musculus</p> <p>GenebankID or UniProtKB</p>
<p>GO:0042803 : protein homodimerization activity (https://www.ebi.ac.uk/QuickGO/term/GO:0042803)</p> <p>GO:0046982 : protein heterodimerization activity (https://www.ebi.ac.uk/QuickGO/term/GO:0046982)</p> <p>GO:0046872 : metal ion binding (https://www.ebi.ac.uk/QuickGO/term/GO:0046872)</p> <p>GO:0004497 : monoxygenase activity</p>		

(<https://www.ebi.ac.uk/QuickGO/term/GO:0004497>)

GO - Biological Process

GO:0032438 : melanosome organization

(<https://www.ebi.ac.uk/QuickGO/term/GO:0032438>)

GO:0043473 : pigmentation (<https://www.ebi.ac.uk/QuickGO/term/GO:0043473>)

GO:0048023 : positive regulation of melanin biosynthetic process

(<https://www.ebi.ac.uk/QuickGO/term/GO:0048023>)

GO:0006583 : melanin biosynthetic process from tyrosine

(<https://www.ebi.ac.uk/QuickGO/term/GO:0006583>)

GO:0030318 : melanocyte differentiation

(<https://www.ebi.ac.uk/QuickGO/term/GO:0030318>)

GO:0043438 : acetoacetic acid metabolic process

(<https://www.ebi.ac.uk/QuickGO/term/GO:0043438>)

GO:0006582 : melanin metabolic process

(<https://www.ebi.ac.uk/QuickGO/term/GO:0006582>)

GO - Cellular Component

GO:0016021 : integral component of membrane

(<https://www.ebi.ac.uk/QuickGO/term/GO:0016021>)

GO:0030669 : clathrin-coated endocytic vesicle membrane

(<https://www.ebi.ac.uk/QuickGO/term/GO:0030669>)

GO:0010008 : endosome membrane

(<https://www.ebi.ac.uk/QuickGO/term/GO:0010008>)

GO:0042470 : melanosome (<https://www.ebi.ac.uk/QuickGO/term/GO:0042470>)

GO:0033162 : melanosome membrane

(<https://www.ebi.ac.uk/QuickGO/term/GO:0033162>)

Presumptive Null

No ([https://www.gephebase.org/search-criteria?/and+Presumptive Null=~No~#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Presumptive+Null=~No~#gephebase-summary-title))

Molecular Type

Coding ([https://www.gephebase.org/search-criteria?/and+Molecular Type=~Coding~#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Molecular+Type=~Coding~#gephebase-summary-title))

Aberration Type

SNP ([https://www.gephebase.org/search-criteria?/and+Aberration Type=~SNP~#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Aberration+Type=~SNP~#gephebase-summary-title))

SNP Coding Change

Nonsynonymous

Molecular Details of the Mutation

c.1487G>A p.Gly496Asp

Experimental Evidence

Candidate Gene ([https://www.gephebase.org/search-criteria?/and+Experimental Evidence=~Candidate Gene~#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Experimental+Evidence=~Candidate+Gene~#gephebase-summary-title))

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	Gly	Asp	496

Main Reference

The brown coat colour of Coppernecked goats is associated with a non-synonymous variant at the TYRP1 locus on chromosome 8. (2015) (<https://pubmed.ncbi.nlm.nih.gov/25392961>)

Authors

Becker D; Otto M; Ammann P; Keller I; DrÄ¼gemÄ¼ller C; Leeb T

Abstract

The recent development of a goat SNP genotyping microarray enables genome-wide association studies in this important livestock species. We investigated the genetic basis of the black and brown coat colour in Valais Blacknecked and Coppernecked goats. A genome-wide association analysis using goat SNP50 BeadChip genotypes of 22 cases and 23 controls allowed us to map the locus for the brown coat colour to goat chromosome 8. The TYRP1 gene is located within the associated chromosomal region, and TYRP1 variants cause similar coat colour phenotypes in different species. We thus considered TYRP1 as a strong positional and functional candidate. We resequenced the caprine TYRP1 gene by Sanger and Illumina sequencing and identified two non-synonymous variants, p.Ile478Thr and p.Gly496Asp, that might have a functional impact on the TYRP1 protein. However, based on the obtained pedigree and genotype data, the brown coat colour in these goats is not due to a single recessive loss-of-function allele. Surprisingly, the genotype distribution and the pedigree data suggest that the (496) Asp allele might possibly act in a dominant manner. The (496) Asp allele was present in 77 of 81 investigated Coppernecked goats and did not occur in black goats. This strongly suggests heterogeneity underlying the brown coat colour in Coppernecked goats. Functional experiments or targeted matings will be required to verify the unexpected preliminary findings.

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Additional References

RELATED GEPHE

Related Genes

3 (Agouti (ASIP), EDNRA, MC1R) ([https://www.gephebase.org/search-criteria?/or+Taxon ID=~9925~/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title](https://www.gephebase.org/search-criteria?/or+Taxon+ID=~9925~/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title))

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

<https://omia.org/OMIA001249/9925/>